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Leukaemia Section
Short Communication

t(16;21)(p11;q22)

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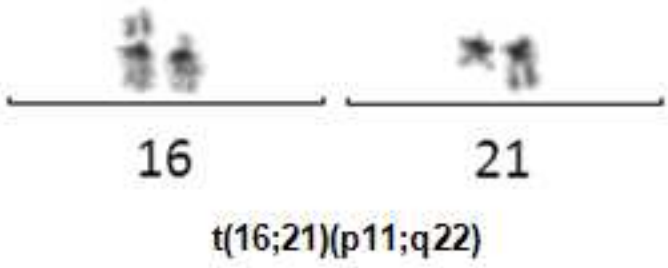

t(16;21)(p11;q22) G- banding - Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease

de novo acute non lymphocytic leukemia (ANLL); to be noted is one case of chronic myelogenous leukemia (CML) -blast crisis.

Phenotype/cell stem origin

ANLL cases: mainly M1, M2, M4, M5a, M5b, or M7 ANLL; may be preceded by a myelodysplastic syndrome (MDS).

Epidemiology

About 40 reported cases, mainly found in young adults; children cases are described; median age is about 30 yrs; balanced sex ratio.

Clinics

Blood data: anemia, thrombocytopenia, mild hyperleucocytosis; with high monocytic cell count at times.

Cytology

Myelocytic and monocytoid features are often present; eosinophils in the bone marrow are sometimes abnormal and/or elevated; erythrophagocytosis may be found.

Prognosis

Seems poor: complete remission may not be achieved; there is high incidence of relapse within a year and a median of survival is about 22 months (cases herein reviewed).

Disease

Ewing tumours

Note

t(16;21)(p11;q22) has been found in rare cases of Ewing tumours, a paediatric neoplasm with small round-cells derived from neural crests cells usually associated with translocations involving EWSR1.

Cytogenetics

Ewing tumours are usually associated with a t(11;22)(q24;q12) with 5' EWSR1 - 3' FLI1 involvement, less often associated with t(21;22)(q22;q12) with 5' EWSR1 - 3' ERG involvement, rarely associated with t(2;22)(q36; q12) (5' EWSR1 - 3' FEV) or with t(17;22)(q21;q12) (5' EWSR1-3' ETV4)

Prognosis

Recent treatments have improved the prognosis of Ewing's tumours. The prognosis is mainly determined by the presence of metastases at the time of diagnosis.

Cytogenetics

Additional anomalies

ANLL cases: found solely in about 60% of cases in at least a subclone; associated with +10, +8, or de(9q)/-9 in about 10% of cases each.

Genes involved and proteins

FUS

Location

16p11

Protein

RNA binding protein; member of the TET family, like EWSR1.

ERG

Location

21q22

Protein

ETS transcription factor.

Result of the chromosomal anomaly

Hybrid gene

Description

5' FUS including exons 1 to 6, 7 or 8 - 3' ERG from exon 7, 8 or 9 to C-term.

Fusion protein

Description

N-term FUS transactivation domain fused to the C-term DNA binding ETS domain of ERG.

Oncogenesis

Seems to act as a transcriptional activator.

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